

Introducing

GENETIC TESTING REGISTRY

<http://www.ncbi.nlm.nih.gov/gtr/>

What is the GTR?

The NIH Genetic Testing Registry (GTR) is a centralized repository of genetic test information voluntarily supplied by test providers. The scope of the Registry includes the purpose, methodology, validity, and evidence of utility of each test, as well as laboratory contacts and credentials. The web site links to context-specific information about conditions, genes, test standards, practice guidelines, and consumer support sites.

The GTR is a test registry, not a laboratory directory. The GTR enables centralized access to detailed information about genetic tests by collecting sufficient information to document the basis and intended purpose(s) of the test, exactly what is measured and how that measurement is done, and the evidence that supports the test's analytical validity, clinical validity, and clinical utility. The web interface allows users to retrieve that information from multiple avenues, including by name of the test, by name of the provider, by category of the test, and by purpose (e.g., name of condition or gene). If a user does a non-specific search (A) for tests (note the menu selection in the query bar present on all pages), and many results are found, the filters in the left column can be used to make selections and quickly focus on the subset of the results of interest. For example, the 1455 results for a query of cardiomyopathy in (A) would quickly be reduced to 17 if Long QT Syndrome 5 were checked in the Apply filters section (B.1). When one disease is selected, then the Laboratory Compare button (boxed in B) can be used to generate a grid (C) summarizing the categories of available tests for that condition from each test provider. Each provider-specific section can be expanded by the control at the right to display more specifics about each category of test (C.1).

The web site provides a portal for medical geneticists.

The GTR web site provides multiple views of medical genetic information: Test details, laboratory summaries, condition-specific reports, and gene-specific reports. This approach makes it easy to display key information, and quickly navigate to the specifics of choice. For example, in (D), which is a page specific to a condition, you see the disease characteristics portion of a *GeneReviews* summary (D.1), with links to the complete *GeneReviews* and each section (D.2); a summary of the tests available (D.3), with links to more details about methodologies; a list of associated genes (D.4); and context-specific links to review articles, practice guidelines, and resources for clinical practice, review of molecular biological details, and consumer resources (D.5).

NCBI Resources (7) How To (2) [Sign Out](#)

GTR: GENETIC TESTING REGISTRY

Conditions/Phenotypes

Noonan syndrome 1

GeneReviews

DISEASE CHARACTERISTICS

Noonan syndrome (NS) is characterized by short stature, congenital heart defect, and developmental delay of variable degree. Other findings can include broad webbed neck, unusual chest shape with superior pectus carinatum and inferior pectus excavatum, cryptorchidism, characteristic facies, valvular conduction defects, lymphatic dysplasias, and ocular abnormalities. Although birth length is usually normal, final adult height approaches the lower limit of normal. Congenital heart disease occurs in 50%-60% of individuals. Pulmonary valve stenosis, often with dysplasia, is the most common heart defect and is found in 20%-50% of individuals. Hypertrophic cardiomyopathy, found in 20%-30% of individuals, may be present at birth or develop in infancy or childhood. Other structural defects include atrial and ventricular septal defects, branch pulmonary artery stenosis, and tetralogy of Fallot. Up to one third of affected individuals have mild intellectual disability. [Full GeneReview](#)

Summary Diagnosis Clinical Description Differential Diagnosis Management Counseling Molecular Genetics Resources References Chapter Notes

Available Tests

53 tests are in the database for this condition. [See all available tests](#)

Tests are available in the following method categories:

- Deletion/duplication analysis (2)
- Mutation screening of select exons (1)
- Sequence analysis of select exons (7)
- Linkage analysis (3)
- Sequence analysis of the entire coding region (37)
- Mutation screening of the entire coding region (2)
- Targeted mutation analysis (1)

Associated Genes

PTN11

Also known as: BPTP3, CFC, MGC14433, NS1, PTP-10, PTP-20, SH-PTP2, SH-PTP3, SHP2

Summary: protein tyrosine phosphatase, non-receptor type 11

Reviews

GeneReviews
Reviews in PubMed
Genetic Testing in PubMed
Diagnosis in PubMed

Clinical Resources

OMIM
ClinGen
Circulatorix.gov

Molecular Resources

OMIM
RefSeqGene
View PTPN11 variations

Consumer Resources

Office of Rare Diseases Research
Genetic Home Reference
MedlinePlus

1 2 3 4 5

cardiomyopathy

GTR Home - Tests - Search results - cardiomyopathy

Apply filters [Reset all](#)

Conditions/Phenotypes

Enter text to filter the list

Select a condition [Reset](#)

Test type [Reset](#)

Test method [Reset](#)

Lab location [Reset](#)

1455 results

HCM Cardiomyopathy

Methods: [Sequence analysis of the entire coding region](#) [Deletion/duplication analysis](#)

Target Population: Patients with clinical features of HCM. Parents, siblings, and possibly children of a patient diagnosed with a mutation in one of the HCM genes. Testing is only performed in a patient with a diagnosis of HCM and has an identified pathogenic gene mutation.

Lab(s): [Dr. Michael J. Ackerman, MD, PhD, Director](#) [Dr. Michael J. Ackerman, MD, PhD, Director](#)

Directors: Dr. Michael J. Ackerman, MD, PhD, Director

Dilated Cardiomyopathy

Methods: [Sequence analysis of the entire coding region](#) [Sequence analysis of select exons](#) [Linkage analysis](#) [Deletion/duplication analysis](#)

Lab(s): [Dr. Michael J. Ackerman, MD, PhD, Director](#)

Directors: Dr. Michael J. Ackerman, MD, PhD, Director

SCN5A-Related Dilated Cardiomyopathy

Methods: [Sequence analysis of the entire coding region](#) [Linkage analysis](#)

Lab(s): [Dr. Michael J. Ackerman, MD, PhD, Director](#)

Directors: Dr. Michael J. Ackerman, MD, PhD, Director

THN12-Related Dilated Cardiomyopathy

Methods: [Sequence analysis of the entire coding region](#) [Deletion/duplication analysis](#)

Lab(s): [Dr. Michael J. Ackerman, MD, PhD, Director](#)

Directors: Dr. Michael J. Ackerman, MD, PhD, Director

THN12-Related Familial Hypertrophic Cardiomyopathy

Methods: [Sequence analysis of the entire coding region](#) [Deletion/duplication analysis](#)

Lab(s): [Dr. Michael J. Ackerman, MD, PhD, Director](#)

Directors: Dr. Michael J. Ackerman, MD, PhD, Director

SCN5A-Related Dilated Cardiomyopathy

Methods: [Sequence analysis of the entire coding region](#) [Deletion/duplication analysis](#)

Lab(s): [Dr. Michael J. Ackerman, MD, PhD, Director](#)

Directors: Dr. Michael J. Ackerman, MD, PhD, Director

PAN1-Related Dilated Cardiomyopathy

Methods: [Sequence analysis of the entire coding region](#) [Deletion/duplication analysis](#)

Lab(s): [Dr. Michael J. Ackerman, MD, PhD, Director](#)

Directors: Dr. Michael J. Ackerman, MD, PhD, Director

long qt syndrome

GTR Home - Tests - Search results - long qt syndrome

Apply filters [Reset all](#)

Conditions/Phenotypes

Enter text to filter the list

Select a condition [Reset](#)

Test type [Reset](#)

Test method [Reset](#)

Lab location [Reset](#)

17 results

Long QT Syndrome 5

Methods: [Sequence analysis of the entire coding region](#) [Deletion/duplication analysis](#)

Lab(s): [Dr. Michael J. Ackerman, MD, PhD, Director](#)

Directors: Dr. Michael J. Ackerman, MD, PhD, Director

Long QT Syndrome 5

Methods: [Sequence analysis of the entire coding region](#) [Linkage analysis](#)

Lab(s): [Dr. Michael J. Ackerman, MD, PhD, Director](#)

Directors: Dr. Michael J. Ackerman, MD, PhD, Director

Long QT Syndrome 5

Methods: [Sequence analysis of the entire coding region](#)

Lab(s): [Dr. Michael J. Ackerman, MD, PhD, Director](#)

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Long QT Syndrome 5

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long qt syndrome

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Directors: Dr. Michael J. Ackerman, MD, PhD, Director

Comparing 10 labs testing Long QT Syndrome 5

Search for:

1 2 3 4 5

Your laboratory in the GTR

Laboratory for Molecular Medicine (Displayed from Gene Tests)

General Information	Conditions and Tests
<p>Laboratory for Molecular Medicine Harvard Medical School and Partners Healthcare, HMS, PHS Cambridge, MA, United States 02139 Website: pcpdm.partners.org/LMM</p>	<p>70 conditions/phenotypes with 70 tests</p> <p>Enter text filters to narrow down the list</p> <ul style="list-style-type: none">3-Methylglutaconic aciduria type 2 1 testAPC-Associated Polyposis Conditions 1 testAlagille syndrome 1 1 testAlpha-B crystallinopathy 1 testAmyloidogenic transthyretin amyloidosis 1 testCardio-facio-cutaneous syndrome 1 testCardio-facio-cutaneous syndrome 1 testCardio-facio-cutaneous syndrome 1 testCardio-facio-cutaneous syndrome 1 testCatecholaminergic polymorphic ventricular tachycardia 1 test <p>70 conditions not shown, use filters above to narrow down the list.</p>
Personnel	List of Services
<p>Director: Heidi L Rehm, PhD, MMSc, FACMG, Laboratory Director</p> <p>Director: Scott Weiss, MD, Laboratory Director</p> <p>Victoria Joshi, PhD, FACMG, Laboratory Associate Director</p> <p>Birgit Funke, PhD, FACMG, Laboratory Associate Director</p> <p>Amy Lovellette Hernandez, MS, CGC, Laboratory Genetic Counselor</p> <p>Samantha Baxter, MS, CGC, Laboratory Genetic Counselor</p> <p>Lab Contact: Priscilla Cepeda, Laboratory Contact</p> <p>Melissa Alford Kelly, MS, CGC, Laboratory Genetic Counselor</p> <p>Katherine Lafferty, MS, Laboratory Genetic Counselor</p>	<p>Ashkenazi Jewish Carrier Test Panel</p> <p>Neonatal/ Costello/ LEOPARD/ Cardofaciocutaneous Syndrome(s) (RAS/MAPK Pathway) Panel</p> <p>Cardiomyopathy (General) Panel</p> <p>Mutation Confirmation</p>
List of Certifications/Licenses	
<p>Certifications</p> <p>CLIA, Number: 22D1005307, Expiration date: 2012-07-01</p>	

Laboratory-specific pages in the GTR display the tests and services offered, as well as how to contact the laboratory. This information is provided by the laboratory.

Laboratories choose which staff members they want to show publicly in the GTR.

Users can bookmark their favorite labs for easy access to the lab's test menu and contact information.

Users can browse the conditions/phenotypes and the associated tests offered by a single laboratory, and view the additional services a laboratory performs.

NCBI Site map All databases Search

Donna Maglott Log out

Your Labs in GTR

Add a new lab Migrate data from GeneTests

Submit test information to the GTR

- Transfer data from GeneTests in one simple click!
- Add, edit and delete your tests online.
- Use GTR's specialized web interface to facilitate submission of supplemental data.
- Take advantage of the bulk upload feature via spreadsheet or XML.

The GTR utilizes the latest software and internet technologies for searching, registration and submission, and provides a range of tools to simplify and speed the process of registering tests. The data entry system has been designed to minimize burden to registrants, with extensive use of menus, "type ahead" functionality, and text fields for those components where submitters might want to cut-and-paste information from their websites and other sources. Where possible, fields are automatically populated for the submitter; for instance, once a submitter fills out the condition for which a test is used, several related fields (e.g., disease identifiers, synonyms, acronyms and disease types) are automatically populated for review. A test copy function is available to expedite entry of information for different tests for which many fields are the same, e.g., if the new test differs in only a few fields such as test name and order code. Submitters can then change any field from the copied test. The GTR also provides a mechanism for bulk submission of data, which will significantly reduce the burden for laboratories that want to provide information on multiple genetic tests or multiplex panel tests.

What information is required?

The GTR is intended to provide information about the test provider as well as the availability, accuracy, validity and usefulness of each test. The complete list of optional (85) and minimal (31) fields being requested is posted on the GTR web site at http://www.ncbi.nlm.nih.gov/projects/gtr/docs/GTR_Proposed_Field_Definitions_distribute.pdf. About half of the minimal fields describe the laboratory (e.g., name, contacts), and are automatically completed after the first entry into the GTR. If a test is migrated from GeneTests, approximately 26 fields are transferred and automatically populated in GTR, leaving only 5 fields to add. For tests being submitted *de novo*, minimal fields are clearly marked for easy entry.

Contact us:

Stay informed of the latest GTR news! Email us at gtr@ncbi.nlm.nih.gov and ask to be subscribed to the GTR listserv. GTR is projected to launch in early 2012. Bookmark NOW! <http://ncbi.nlm.nih.gov/gtr>

